



SLC30A10 gene

solute carrier family 30 member 10

Normal Function

The *SLC30A10* gene provides instructions for making a protein that transports the element manganese across cell membranes. Manganese is important for many cellular functions, but large amounts are toxic, particularly to brain and liver cells. The *SLC30A10* protein is found in the membranes surrounding liver cells and nerve cells in the brain, as well as in the membranes of structures within these cells. The protein protects these cells from high concentrations of manganese by removing manganese when levels become elevated.

Health Conditions Related to Genetic Changes

hypermanganesemia with dystonia, polycythemia, and cirrhosis

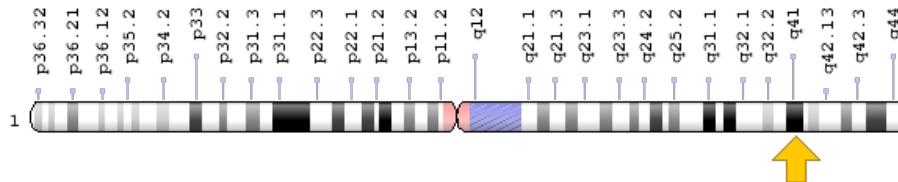
Several mutations in the *SLC30A10* gene have been identified in people with hypermanganesemia with dystonia, polycythemia, and cirrhosis (HMDPC). This inherited disorder is characterized by high levels of manganese in the blood (hypermanganesemia). The disorder causes movement problems, such as involuntary tensing of the muscles (dystonia); an increased number of red blood cells (polycythemia); and liver abnormalities, including liver disease (cirrhosis).

Mutations in the *SLC30A10* gene impair the transport of manganese out of the cell, allowing the element to build up in brain and liver cells. Manganese accumulates in a region of the brain that helps control movement, damaging nerve cells and leading to the movement problems characteristic of HMDPC. Damage caused by buildup of manganese in the liver causes the characteristic liver problems. High levels of manganese help increase the production of red blood cells, so excess amounts of this element also result in polycythemia.

Chromosomal Location

Cytogenetic Location: 1q41, which is the long (q) arm of chromosome 1 at position 41

Molecular Location: base pairs 219,910,398 to 219,959,754 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DKFZp547M236
- HMDPC
- solute carrier family 30, member 10
- zinc transporter 10
- ZnT-10
- ZNT10
- ZNT10_HUMAN
- ZRC1

Additional Information & Resources

Educational Resources

- Agency for Toxic Substances and Disease Registry: Manganese
<https://www.atsdr.cdc.gov/substances/toxsubstance.asp?toxicid=23>
- Oregon State University Linus Pauling Institute: Manganese
<http://lpi.oregonstate.edu/mic/minerals/manganese>

GeneReviews

- Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease
<https://www.ncbi.nlm.nih.gov/books/NBK100241>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC30A10%5BTIAB%5D%29+OR+%28ZRC1%5BTIAB%5D%29+OR+%28ZNT10%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 30 (ZINC TRANSPORTER), MEMBER 10
<http://omim.org/entry/611146>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC30A10.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC30A10%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25355
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/55532>
- UniProt
<http://www.uniprot.org/uniprot/Q6XR72>

Sources for This Summary

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